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What is claimed is:

1. Isolated nucleic acid encoding a human EHOC-1 polypeptide.

2. Isolated nucleic acid according to claim 1, wherein said nucleic acid comprises DNA.

3. DNA according to claim 2, wherein said DNA is a cDNA.

4. DNA according to claim 2, wherein said DNA encodes the amino acid sequence set forth in SEQ ID NO: 2.

5. DNA according to claim 2, wherein said DNA hybridizes under high stringency conditions to substantially the entire coding sequence (nucleotides 157-3726) set forth in SEQ ID NO: 2.

6. DNA according to claim 2, wherein said DNA has substantially the same nucleotide sequence as the nucleotide sequence set forth in SEQ ID NO: 1.

7. A vector comprising DNA according to claim 2.

8. A host cell containing a vector according to claim 7, wherein said cell is a procaryotic cell or a eucaryotic cell.

9. A host cell according to claim 8, wherein said cell expresses a functional EHOC-1 protein.

10. A nucleic acid probe comprising at least 15 nucleotides capable of specifically hybridizing with a sequence of nucleic acids of the nucleotide sequence set forth in SEQ ID NO: 1.

11. A nucleic acid probe according to claim 10, wherein said probe is labeled with a detectable marker.

12. A kit for detecting mutations and aneuploidies in chromosome 21 at locus q22.3 comprising a plurality of probes, wherein each probe comprises a nucleic acid sequence having at least 15 bp of contiguous nucleotides capable of specifically hybridizing with a sequence of nucleic acids of the nucleotide sequence set forth in SEQ ID NO: 1, and wherein each individual probe corresponds to a specific locus on chromosome 21q22.3

13. Isolated mRNA complementary to DNA according to claim 2.

14. An oligonucleotide composition comprising chemical analogues of the nucleic acid of claim 2 operatively linked to a promoter of RNA transcription.

15. An antisense oligonucleotide capable of specifically binding to and modulating the translation of mRNA according to claim 13.

16. Isolated EHOC-1 polypeptide and functional equivalents thereof.

17. Isolated EHOC-1 polypeptide according to claim 16, wherein said polypeptide has substantially the same amino acid sequence as that set forth in SEQ ID NO: 2.

18. Isolated EHOC-1 polypeptide according to claim 16, wherein said polypeptide has the same amino acid sequence as that set forth in SEQ ID NO: 2.

19. Isolated EHOC-1 polypeptide according to claim 16, wherein said polypeptide is encoded by a

nucleotide sequence that is substantially the same nucleotide sequence as that set forth in SEQ ID NO: 1.

20. Isolated EHOC-1 polypeptide according to claim 16, wherein said polypeptide is encoded by the nucleotide sequence set forth in SEQ ID NO: 1.

21. An EHOC-1 polypeptide expressed recombinantly in a host cell.

22. An EHOC-1 polypeptide according to claim 21, wherein said polypeptide is encoded by a nucleotide sequence that is substantially the same as the nucleotide sequence set forth in SEQ ID NO: 1.

23. An EHOC-1 polypeptide according to claim 21, wherein said polypeptide is encoded by the nucleotide sequence set forth in SEQ ID NO: 1.

24. An antibody that specifically binds to a determinant on a human EHOC-1 protein or active fragment thereof.

25. An antibody according to claim 24, wherein said antibody is a monoclonal antibody.

26. An antibody according to claim 24, wherein said antibody is a polyclonal antibody.

27. A composition comprising an amount of the antisense oligonucleotide according to claim 13 effective to modulate expression of a human EHOC-1 polypeptide and an acceptable hydrophobic carrier capable of passing through a cell membrane.

28. A composition according to claim 27, wherein the oligonucleotide is coupled to a substance which inactivates mRNA.

29. A composition according to claim 28, wherein said substance is a ribozyme.

30. A composition comprising an amount of an antibody according to claim 24 effective to block binding of naturally occurring ligands to the human EHOC-1 receptor and an acceptable carrier.

31. A transgenic nonhuman mammal expressing DNA encoding a human EHOC-1 polypeptide.

32. A transgenic nonhuman mammal according to claim 31, wherein said DNA encoding said polypeptide has been mutated as to be incapable of normal polypeptide activity, and wherein the polypeptide so expressed is not native EHOC-1 polypeptide.

33. A transgenic nonhuman mammal, the genome of which comprising antisense DNA complementary to DNA encoding a human EHOC-1 polypeptide, wherein said antisense DNA is transcribed into antisense mRNA complementary to mRNA encoding a human EHOC-1 polypeptide.

34. A transgenic nonhuman mammal according to claim 31, wherein said DNA is operatively linked to an inducible promoter.

35. A transgenic nonhuman mammal according to claim 31, wherein said DNA is operatively linked to tissue specific regulatory elements.

36. A transgenic nonhuman mammal according to claim 31, wherein the transgenic nonhuman mammal is a mouse.

37. A method for identifying nucleic acids encoding a human EHOC-1 protein, said method comprising:

contacting a sample containing nucleic acids with a probe according to claim 11, wherein said contacting is effected under high stringency hybridization conditions, and identifying compounds which hybridize thereto.

38. A method for identifying compound(s) which bind to a human EHOC-1 polypeptide, said method comprising contacting cells according to claim 9 with said compound(s) and identifying compounds which bind thereto.

39. A method for detecting the presence of a human EHOC-1 polypeptide on a cell surface, said method comprising contacting a test cell with an antibody according to claim 24, detecting the presence of an antibody-receptor complex, and therefor detecting the presence of a human EHOC-1 polypeptide on the cell surface.

40. A method for diagnosing a predisposition to a disorder associated with the expression of a specific human EHOC-1 polypeptide allele, said method comprising:

contacting a sample containing nucleic acids with a plurality of probes, wherein each probe comprises a nucleic acid sequence having at least 15 bp of contiguous nucleotides capable of specifically hybridizing with a sequence of nucleic acids of the nucleotide sequence set forth in SEQ ID NO: 1, and wherein each individual probe corresponds to a specific locus on chromosome 21q22.3

41. A method according to claim 40, wherein said disorder is selected from progressive myoclonus epilepsy, holoprosencephaly, or autoimmune polyglandular disease.

42. A method for deterring the onset of symptoms associated with particular disorder comprising

administering a composition which modulates expression of gene.

43. A method for introducing changes at human chromosome locus 21q22.3 comprising transforming a sample of cells obtained from a subject having progressive myoclonus epilepsy with the nucleic acid according to claim 1 along with a selective marker gene; maintaining cells in selective media; and

isolating viable cells containing a modified target sequence.

44. A method of supplying wild-type EHOC-1 gene function to a cell which has a mutation/aneuploidy in the EHOC-1 gene comprising introducing a wild-type EHOC-1 gene or functional fragment thereof into said cell such that it is expressed.

45. Single strand DNA primers for amplification diagnosis of progressive myoclonus epilepsy, wherein said primers comprise a nucleic acid sequence derived from the nucleic acid sequence set forth as SEQ ID NO: 1.

46. A method for detecting one or more EHOC-1 alleles in a sample of nucleic acid comprising determining the presence or absence of variant nucleotide sequence in a gene contained in any of BAC clones.